Diagnostic Ophthalmology
Ophtalmologie diagnostique

Bianca S. Bauer, Lynne S. Sandmeyer, Bruce H. Grahn

History and clinical signs

An 18-month-old intact male Australian shepherd dog was referred to the ophthalmology service at the Western College of Veterinary Medicine for an abnormally appearing left optic nerve noted on routine indirect ophthalmoscopic evaluation by the referring veterinarian. Vaccinations for distemper, parvo, and rabies viruses were current and the dog had recently been dewormed. Neuro-ophthalmic examination was within normal limit bilaterally. Schirmer tear test (Schirmer Tear Test Strips; Alcon Canada, Mississauga, Ontario) values were 20 mm/min bilaterally. The intraocular pressures were estimated with a rebound tonometer (Tonovet; Tiolat Oy, Helsinki, Finland) and were 15 mmHg bilaterally. Biomicroscopic (Osram 64222; Carl Zeiss Canada, Don Mills, Ontario) and indirect ophthalmoscopic (Heine Omega 200; Heine Instruments Canada, Kitchener, Ontario) examinations were completed. Abnormalities were not detected on biomicroscopic examination; however, indirect ophthalmoscopic examination revealed an abnormal left optic nerve head with a deep cavitation (Figure 1).

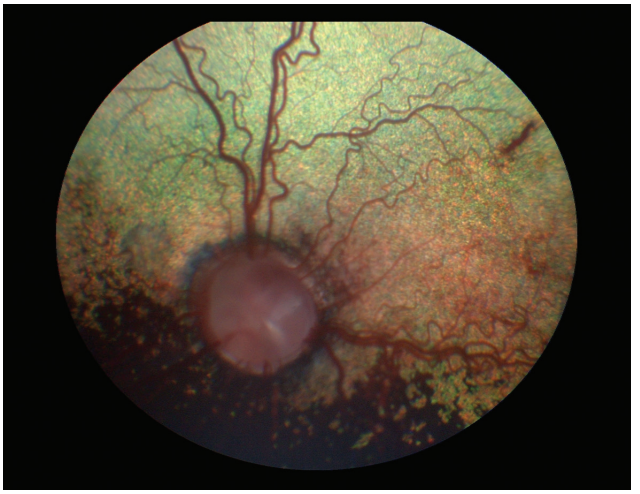

Figure 1. Fundus photograph of the left eye of an 18-month-old Australian shepherd dog.

What are your clinical diagnoses, differential diagnoses, diagnostic plan, and prognosis?

Discussion

Our clinical diagnosis was a large left optic nerve head coloboma involving the entire diameter of the optic nerve head. A coloboma is an absence of tissue that is normally present. Colobomata are congenital and non-progressive; they may affect any ocular tissue. Typical colobomata are caused by faulty closure or fusion of the ventral embryonic fissure, while atypical colobomata develop due to lack of induction or impaired proliferation of the affected tissues (1). Colobomata frequently involve the optic disc or the immediate region around the disc but they may also be located in the peripheral fundus (2). A coloboma manifests as a sharply circumscribed, usually pale, depigmented area with reduced or anomalous vasculature and is often cupped. Inherited colobomatous defects are found in albino herefords and Charolais cattle (3,4), collie eye anomaly (5), Australian shepherd dogs (6), and basenji dogs (7).

Collie eye anomaly (CEA) is a congenital, non-progressive, ocular disease with variable manifestations (4). The condition involves defects of the posterior vascular and fibrous tunics of the eye and the pathogenesis is considered to be abnormal mesodermal differentiation resulting in defects mainly of the sclera, choroid, optic disc, retina, and retinal vasculature (8). The CEA locus is mapped to a 3.9-cM region of chromosome 37 (9). Collie eye anomaly is very common and widespread geographically in the collie breeds (smooth and rough collie, border collie, and Shetland sheepdog) and has also been described in various non-collie breeds (Australian shepherd, beagle, dachshund, German shepherd dog, Siberian husky) as well as a mixed breed dog (4,10–12). In the collie, the condition has been considered to be due to a simple recessive gene independent of gender or coat color (13). Collie eye anomaly is bilateral, although not necessarily symmetrical between eyes, and the severity of the disease ranges from no apparent visual deficits to total blindness (8). Bilateral chorioretinal hypoplasia is considered the diagnostic lesion for CEA. These variably sized, pale regions are located temporal to the optic discs and they may or may not be accompanied by other defects that are included in the syndrome (colobomata, retinal detachments, or intraocular hemorrhage).

Optic nerve head (ONH) colobomata may appear as enlarged discs with a deep cavitation or as slightly enlarged, irregular discs

Department of Small Animal Clinical Sciences, Western College of Veterinary Medicine, University of Saskatchewan, 52 Campus Drive, Saskatoon, Saskatchewan S7N 5B4.

containing deep pits within the borders of the ONH. These colobomata may be "typical" at the 6 o'clock position or "atypical" at the nasal or temporal disc margin and they are always associated with varying degrees of choroidal hypoplasia (11). Optic disc colobomata may be small and hard to differentiate from a deep physiologic cup, or they may be very large and deep such that they distort the area of the disc severely. Large ONH colobomata can be easily identified by the color of the lesion and the way the retinal vasculature changes course or disappears at the rim of the defect. Differential diagnoses for a large ONH coloboma include optic nerve head cupping or atrophy due to glaucoma or trauma. In this case, however, the dog's age, breed, and overall clinical findings ruled out pathologic ONH cupping. Despite their frequent dramatic appearance, little or no visual deficits are noted with ONH colobomata (14).

Collie eye anomaly is most accurately diagnosed ophthalmoscopically at 6 to 7 wk of age (8). A commercial mutation detection test (OptiGen® LLC; Cornell Business & Technology Park, Ithaca, New York, USA) is also available to identify the genetic status of breeding animals. In this particular case, submission of blood for DNA-based testing was warranted and was performed to confirm CEA genotype. Blood testing by OptiGen® confirmed an affected CEA genotype. Treatment for CEA is usually not necessary and no treatment was performed in this case. If present, focal retinal detachments associated with colobomata can be treated with laser photocoagulation (15). The long-term prognosis for this dog is good; however, should this dog be used for breeding, it is recommended that he only be bred to a mate that is genotyped as homozygous normal to avoid the production of CEA-affected puppies.

References

1. Grahn BH, Peiffer RL. Veterinary ophthalmic pathology. In: Gelatt KN, Gilger BC, Kern TJ, eds. Veterinary Ophthalmology. 5 ed. Vol. I. Ames, Iowa: John Wiley & Sons, 2013:435–523.
2. Martin CL. Ophthalmic Disease in Veterinary Medicine. London, UK: Manson Publishing, 2005.
3. Gelatt KN, Huston K, Leipold HW. Ocular anomalies of incomplete albino cattle: Ophthalmoscopic examination. Am J Vet Res 1969;30:1313–1316.
4. Barnett KC, Ogien AL. Ocular colobomata in Charolais cattle. Vet Rec 1972;91:592.
5. Roberts SR. The collie eye anomaly. J Am Vet Med Assoc 1969;155:859–864.
6. Gelatt KN, Veith LA. Hereditary multiple ocular anomalies in Australian shepherd dogs. (preliminary report). Vet Med Small Anim Clin 1970;65:39–42.
7. Rubin LF. Inherited Eye Diseases in Purebred Dogs. Baltimore, Maryland: Williams & Wilkins, 1989.
8. Narfstrom K, Petersen-Jones S. Diseases of the canine ocular fundus. In: Gelatt KN, ed. Veterinary Ophthalmology. 4 ed. Vol. 2. Ames, Iowa: Blackwell Publishing, 2007:944–1025.
9. Parker HG, Kukekova AV, Akey DT, et al. Breed relationships facilitate fine-mapping studies: A 7.8-kb deletion cosegregates with Collie eye anomaly across multiple dog breeds. Genome Res 2007;17:1562–1571.
10. Rubin LF, Nelson EJ, Sharp CA. Collie eye anomaly in Australian Shepherd dogs. Prog Vet Comp Ophthalmol 1991;1:105–108.
11. Rubin LF. Inherited Eye Diseases in Purebred Dogs. Philadelphia, Pennsylvania: Lipponcott Williams & Wilkins, 1989.
12. Rampazzo A, D'Angelo A, Capucchio MT, Sereno S, Peruccio C. Collie eye anomaly in a mixed-breed dog. Vet Ophthalmol 2005;8:357–360.
13. Yakely WL, Wyman M, Donovan EF, Fechheimer NS. Genetic transmission of an ocular fundus anomaly in Collies. J Am Vet Med Assoc 1968;152:457–461.
14. Narfstrom K, Petersen Jones SM. Diseases of the canine ocular fundus. In: Gelatt KN, Gilger BC, Kern TJ, eds. Veterinary Ophthalmology. 5th ed. Vol. 2. Ames, Iowa: John Wiley & Sons, 2013:1303–1392.
15. Vainisi SJ, Peyman GA, Wolf ED, West CS. Treatment of serous retinal detachments associated with optic disk pits in dogs. J Am Vet Med Assoc 1989;195:1233–1236.